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JIA-LIKE IN A BOY WITH ATAXIA-TELANGIECTASIA

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Introduction

Ataxia-Telangiectasia (A-T) is an autosomal recessive disorder, caused by mutations in the ATM-gene leading to misfunction of ATM, a protein kinase that is involved in double stand breaks-damage-sensing and cell-cycle-arrest, crucial for DNA-repair. Cells with higher turn-over rates such as neurological and immune cells, are more susceptible to DNA damage, rendering A-T patients to a degenerative, progressive disease consisting in neurological impairment, increased susceptibility to malignancy and predisposition to infections. Despite being linked with many primary immunodeficiencies (PID), juvenile idiopathic arthritis (JIA) has rarely been associated with DNA-repair disorders, such as A-T.

We report a case of JIA-like in a male patient with A-T.

Case Report

Birth ♂, second child of healthy, unrelated parents. Brother 4 years older diagnosed with Ataxia-Telangiectasia at 6 years-old.

Healthy during the first year of life

1 year Stars walking – **unstable gait**

Two disease associated mutations of ATM gene

C.3894 dupt-exon 26
C.5291T>C-exon 35

→ **Ataxia-Telangiectasia**

2 years **Primary immunodeficiencies**

Table 1 – Immunologic evaluation

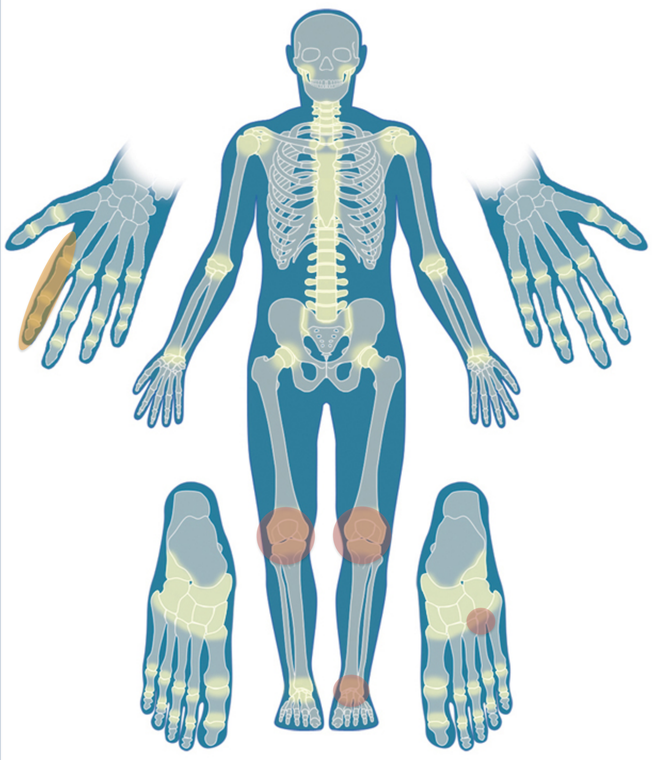
	At diagnosis (2.5 YO)	5 YO	Normal values
IgA (g/L)	**<0,23**	**<0,23**	0,33-3,60
IgM	1,56	1,74	0,55-2,10
IgG	11,30	10,2	5,93-17,30
IgE	<2,0	<2,0	
Lymphocytes (/mm3)	**1870**	**1740**	3000-9500
CD3+ (cells/µl)	**545**	**485**	1400-3700
CD4+ (%CD4CD45RA)	**300 (7%)**	**274 (9%)**	700-2200
CD8+ (%CD8CD45RACD62l)	**131 (28%)**	**133 (40%)**	490-1300
CD19+	**183**	**154**	390-1400
NK-cells (cells/µl)	348	386	130-720
CD3+TCRγδ (%)	**144 (29%)**	**170 (30%)**	1-10%
Vbeta repertoire: oligoclonal, restricted			
Abnormal proliferative lymphocyte response to mitogens and recall Antigens			

- ✓ CD4 and CD8 naif lymphopenia
- ✓ Abnormal proliferative responses
- ✓ Skewed Vbeta repertoire
- ✓ Relative NK lymphocytosis
- ✓ Expanded gammadelta T cells

3 years **Pain** and **swelling** of 4th metatarsophalangeal (MTP) joint of left foot

4 years After 6 months **symptoms persisted**

Rheumatology



Arthritis of:
- Both knees
- Left ankle
- 4th MTP of the left foot

Tenosynovitis of:
- 2nd finger of the right hand

→ **JIA-like**

Ibuprofen (30 mg/kg/day)
Chemical synovectomy with triamcinolone hexacetonide (TH) (knees and ankle)

Remain active:
Arthritis of 4th MTP of left foot
Tenosynovitis of 2nd finger of right hand

Table 2 – Laboratory investigations

Erythrocyte sedimentation rate	**25 mm/h**
C- reactive protein	1,45 mg/L
Antinuclear antibodies	**positive (1/80)**
Anti-cyclic citrullinated protein	negative
Rheumatoid factor	negative

Terapheutic ??? ✗ **Methotrexate?**

Hydroxichloroquine (5 mg/kg/day)

Conclusion

This case seems to be the first known pediatric patient with A-T who developed chronic, JIA-like oligoarthritis. The management of these patients is particularly difficult because they are extremely susceptible to DNA damage and show an increased susceptibility to viral infections (namely herpetic). These features are very important when considering the best therapeutic options for JIA-like arthritis.

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